Why do doctors ask for genetic tests in Autism like condition?

In summary, the literature indicates that the following approximate diagnostic yields are expected in the genetic evaluation of ASDs:

- CMA (10%)
- Fragile X (1–5%)
- *MECP2* (4% of females)
- *PTEN* (5% of those with head circumferences >2.5 SDs that are tested)
- Karyotype (3%)
- Other (10%). Currently, there are no published studies that collate the yield on the other identifiable etiologies of autism. As noted above, identifiable brain anomalies, genetic syndromes, metabolic disorders, and other diagnosable conditions will be identified in the genetic evaluation of persons with ASDs. Using empiric estimates and clinical experience, this has been estimated as 10%.[74]

Therefore, using current knowledge and technology, a thorough clinical genetics evaluation of patients with ASDs is estimated to result in an identified etiology in 30–40% of individuals.

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Table 2 Recently described metabolic conditions associated with an ASD phenotype

3β-Hydroxycholesterol-7-reductase deficiency (Smith–Lemli–Opitz syndrome)
6-*N*-trimethyllysine dioxygenase deficiency
Adenylosuccinate lyase deficiency
Cerebral folate deficiency
Cytosolic 5′ nucleotidase superactivity
Dihydropyrimidinase deficiency
Disorders of creatine transport or metabolism
Disorders of γ-aminobutyric acid metabolism
Phosphoribosylpyrophosphate synthetase superactivity
Succinic semialdehyde dehydrogenase deficiency
Sulfation defects

ASD, autism spectrum disorder.

THIS IS ONE OF COMMON REASONS **WHY ALL AUTISM DIAGNOSED BY PSYCHOLOGISTS OR THERAPISTS ARE NOT ALWAYS AUTISM**.
ALSO THE REASON WHY SOME AUTISMS **DONOT IMPROVE DESPITE THERAPY, AND MAY NEED MEDICINES OF TREATMENT AS PER RESPECTIVE ISSUES IN NEUROLOGICAL OR GENETIC PROBLEM.**
THIS IS BASED ON ACMG GUIDELINES

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Table 5 Selected genetic syndromes that are known etiologies of autism spectrum disorders

22q11.2 deletions including velocardiofacial (Shprintzen) syndrome
Angelman syndrome
CHARGE syndrome
de Lange syndrome
Fragile X syndrome
MED12 disorders (including Lujan–Fryns syndrome)
Prader–Willi syndrome
PTEN-associated disorders (Cowden syndrome, Bannayan–Riley–Ruvalcaba syndrome)
Rett syndrome
Smith–Lemli–Opitz syndrome
Smith–Magenis syndrome
Sotos syndrome
Tuberous sclerosis

PTEN, phosphatase and tensin homolog.

Adapted from ref. 74.

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Table 3 Clinical symptoms that prompt metabolic or mitochondrial testing in persons with ASDs

Acid/base or electrolyte disturbances
Anemia with an elevated mean corpuscular volume
Cyclic vomiting
Dermatologic changes: alopecia, hypertrichosis, and pigmented skin eruptions
Developmental regression associated with illness or fever
Gastrointestinal dysfunction, gastroparesis
Hypotonia/dystonia
Lactic acidosis
Lethargy
Multisystem involvement, especially cardiac, hepatic, or renal (physical and/or laboratory evidence)
Neurodegeneration outside of the typical ASD speech loss at 18–24 months
Poor growth, microcephaly
Seizures

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Table 1 Selected genetic "hot spots" in autism as identified by selected testing modalities

Region	Linkage	Cytogenetics	Microarray	Genomic coordinates	Deletion/ duplication	Candidate genes in the region
1p			1p36.13	1:762,978–17,148,920	dup	
1q			1q21.1	1:145.0–146.4 Mb	dup	*HYDIN*
	1q41–42		1q41	1:214,500,000–236,600,000	del	*MARK1*
2p		2p16	2p16.3	2:50,145,642–51,259,673	del	*NRXN1*
2q	2q		2q31.1	2:93,300,000–243,199,373	del, dup	*SLC25A12*
		2q37	2q37	2:231,972,946–231,989,823	del	*5-HTR2B*
3q			3q24	3:142,984,063–143,567,372	del, dup	*SLC9A9*
	3q25–27			3:148,900,000–187,900,000		
5p			5p15.1	5: 9,623,122–9,624,122	del	
7q			7q11.23	7:72,200,000 (1.5 Mb)	dup	
	7q 22–31	7q	7q22.1	7:98,000,000–107,400,000	del	*RELN*
			7q31.2	7:116,312,458–116,438,439	del, dup	*FOXP2, WNT2, MET*
			7q35–q36	7:145,813,452–148,118,089	del	*EN2, CNTNAP2*
11q			11q13.3–q13.4	11:70,313,960–70,935,807	del	*SHANK2*
12q	12q14.2			12:63,100,000–65,100,000		
13q	13q		13q14.2–q14.1	13:47,300,000–47,300,000	del	*5-HTR2A*
15q 11–13	15q 11–13	15q11–13	15q11–13	15: 20.7–26.7 (Mb)	dup	*UBE3A, SNRPN, CHRNA7*
			15q13.3	15: 28,736,917–30,686,830	del	*CHRNA7*
16p	16p		16p11.2	16:29.5–30.1 (Mb)	del, dup	
			16p13.11	16:15.5–16.5 (Mb)	del, dup	
17p			17p11.2	17:20,156,497–22,975,771	del, dup	
17q	17q11–12		17q11.2	17:24,000,000–31,800,000	del	*SLC6A4*
			17q21.3	17:38,100,000–50,200,000	del, dup	*ITGB3*
18q	18q21–23	18q	18q21.1	18:47,793,251–47,808,143	del	*TCF4, MBD1*
21p	21p13–q11			21:0–16,400,000		
22q		22q11–13	22q11.2	22:19,744,225 (1.5–3.0 Mb)	del	*CRKL, FGF8, TBX1*
			22q13.3	22:51,113,069–51,171,639	del	*SHANK3*
Xp	Xp22	Xp	Xp22.31	X:6,463,313–8,091,810	del, dup	*NLGN4*
Xq	Xq13		Xq13.1	X:70,364,680–70,391,050	del, dup	*NLGN3*
			Xq28	X:153,287,263–153,363,187	del	*MECP2*

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Not all autism are genetic as the genetic database is not yet completely studied and also issues caused by some insult or damage to brain in minor way can also cause autism like developmental disorders.

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